

Client: Example Client ABC123
123 Test Drive
Salt Lake City, UT 84108
UNITED STATES

Physician: Doctor, Example

Patient: Patient, Example

DOB: 3/26/1974
Gender: Male
Patient Identifiers: 01234567890ABCD, 012345
Visit Number (FIN): 01234567890ABCD
Collection Date: [REDACTED]

Achondroplasia (FGFR3) 2 Mutations

ARUP test code 0051266

Achondroplasia PCR Specimen whole Blood

Achondroplasia PCR Negative

Indication for testing: Rule out achondroplasia.

Negative: The fibroblast growth factor receptor (FGFR) 3 gene mutations, G1138A and G1138C, were not present. Therefore this individual is unlikely to be affected with achondroplasia. Rarely, an affected person may have another mutation not detectable by this assay.

This result has been reviewed and approved by Rong Mao, M.D.

BACKGROUND INFORMATION: Achondroplasia (FGFR3) 2 Mutations

CHARACTERISTICS: Short stature with disproportionately short arms and legs, a large head, usually normal life span and intelligence; increased risk for death in infancy from compression of spinal cord and/or upper airway obstruction.
INCIDENCE: 1:25,000
INHERITANCE: Autosomal dominant; 80 percent arise from de novo mutations.
PENETRANCE: 100 percent
CAUSE: Pathogenic FGFR3 gene mutation
CLINICAL SENSITIVITY: Two mutations, c.1138G>A and c.1138G>C, in the FGFR3 gene account for greater than 99 percent of cases.
METHODOLOGY: PCR and fluorescent hybridization probes.
ANALYTICAL SENSITIVITY AND SPECIFICITY: Greater than 99 percent.
LIMITATIONS: Mutations other than c.1138G>A and c.1138G>C will not be detected. Diagnostic errors can occur due to rare sequence variations.

Test developed and characteristics determined by ARUP Laboratories. See Compliance Statement C: aruplab.com/CS

H=High, L=Low, *=Abnormal, C=Critical

VERIFIED/REPORTED DATES

Procedure	Accession	Collected	Received	Verified/Reported
Achondroplasia PCR Specimen	18-132-400275	5/11/2018 8:53:00 AM	5/12/2018 6:44:29 AM	5/15/2018 9:19:00 AM
Achondroplasia PCR	18-132-400275	5/11/2018 8:53:00 AM	5/12/2018 6:44:29 AM	5/15/2018 9:19:00 AM

END OF CHART

H=High, L=Low, *=Abnormal, C=Critical